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CASE REPORT

Multiple Congenital Anomalies in a Preterm Neonate with G6PD Deficiency from Consanguineous Parents, a Case Report

Running Title: Multiple Congenital Anomalies and G6PD Deficiency

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ABSTRACT

Introduction: Congenital anomalies can be defined as structural or functional disorders, including metabolic disorders. The prevalence of congenital anomalies is not high. **Case Presentation:** We would like to present a 34-week- preterm neonate with glucose-6-phosphate dehydrogenase deficiency who was born with several fetal anomalies from consanguineous parents. Mother had a twin-birth pregnancy which one of twins died because of lung hemorrhage and the second twin was born with multiple anomalies four fingers in both hands, short legs attached to the pelvic bone, and absence of tibia, fibula, and ankle bones. Hemimelia, which was mostly known as the congenital deformity and a type of phocomelia, is extremely rare. To detect most of these types of anomalies, magnetic resonance imaging, radiography, ultrasound, and computed tomography scan can be used. **Conclusion:** Congenital anomalies are the cause of many infants' deaths. To detect most of these types of anomalies, MRI, radiography, ultrasound, and computed tomography (CT) scan can be used
Keywords: *Multiple Congenital Anomalies, Glucose-6-phosphate Dehydrogenase (G6PD) Deficiency, Hemimelia*

INTRODUCTION

Congenital anomalies (CA) can be defined as structural or functional disorders, including metabolic disorders, at birth, which is the cause of many infants' deaths. Approximately 70% of infants die in the first month (1, 2). These abnormalities includes cleft lip and cleft palate, cerebral palsy, fragile X syndrome, down syndrome, spina bifida, cystic fibrosis, hydrocephaly, anencephaly, craniorachischisis, encephalocele, gastroschisis and reduction defects of upper and lower limbs. The prevalence of these diseases is not high, and according to the World Health Organization WHO occur in about 3% of infants, which varies from country to country. In general, CA is more likely to occur in males than females (3, 4). A study in North West of Iran on CA types showed a high incidence of anencephaly, hydrocephaly and cleft palate without cleft lip and a small amount for spina bifida and a very high rate for limb reduction defects (5).

The causes of fetal and congenital abnormalities are 30-40% of the genetic causes and approximately 5-10% belong to the environmental factors. Among the genetic causes, monogenous disorders, and chromosomal abnormalities are the most frequent respectively, however, the cause of 40-60% of congenital anomalies is still unknown (2).

Hemimelia, which was mostly known as the congenital deformity and a type of phocomelia, is extremely rare. The most common defects in the lower limbs are Hemimelia fibular, Hemimelia tibial and Hemimelia femoral (6).

In general, effective factors can include genetic disorders, socioeconomic factors, and nutritional factors such as maternal obesity, pregnancy infections, and use of certain drugs, ionizing radiation, and chemicals, as well as air pollution. Different pregnancy conditions such as gestational diabetes mellitus (7), high blood pressure during pregnancy such as antepartum hemorrhage, twin pregnancies, oligohydramnios and, polyhydramnios are also associated with various CA abnormalities.

Glucose and phosphate dehydrogenase deficiency (G6PD) is one of the enzyme disorder in humans that most often affects males. It happens when the body does not have enough of an enzyme called glucose-6-phosphate dehydrogenase (G6PD). the human without G6PD are sensitive to some medicines, foods, and infections and causes a spectrum of disease including neonatal hyperbilirubinemia, acute hemolysis, and chronic hemolysis (8, 9).

Previous research has also shown that family marriages increase the probability of CA (1, 10-12). Here we would like to report a case of preterm neonate from consanguineous parents with multiple congenital anomalies including four fingers in both hands, short legs attached to the pelvic bone, and absence of tibia, fibula, and ankle bones.

CASE PRESENTATION

A 34-week preterm neonate was born with several fetal anomalies from consanguineous parents. Mother (26 years old) had a twin-birth pregnancy which one of twins died because of lung hemorrhage despite resuscitation in neonatal intensive care unit (NICU) and the second twin was born through cesarean section with multiple anomalies. This birth occurred in Shohada of Behshahr Hospital, Mazandaran University of Medical Sciences, Behshahr, Iran.

This neonate was the second child of the family. Checking the maternal health status showed that she had hypothyroidism and malnutrition, which was abandoned without treatment due to the financial disability of the family. The mother did not have prenatal care during pregnancy due to financial problems. In physical examination, there was a soft round mass, approximately 5 cm x 5 cm with defined border in the left frontal part of the head overcoming on the left eye. There were four fingers in both hands and short legs attached to the pelvic bone (figure 1). In postnatal follow-up, magnetic resonance imaging (MRI), radiography, and ultrasound were performed.

Figure 1. A 34-week preterm neonate (23-day-old baby) with congenital anomalies including frontal mass (5 x 5 cm), four fingers in both hands, short legs attached to the pelvic bone, and absence of tibia, fibula, and ankle bones

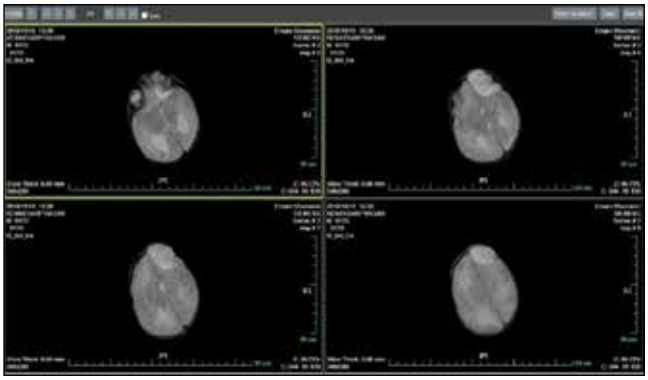

Figure 2. Brain MRI of the neonate from well-defined fluid-filled mass in the left frontal part of the head overcoming on the left eye was suggested to occur because of be left frontal lobe atrophy

The findings of radiography revealed malformation as the absence of tibia, fibula, and ankle bones. In ultrasonography imaging of the frontal area and left orbital, expansion of extra-axial space with an internal echo around the atrophied brain tissue was suggestive to be meningocele or cephalocele. Therefore, MRI was suggested for closer investigation of the left frontal tissue. Results showed the tissue was the part of neonate's brain and expanded because of left frontal lobe atrophy (figure 2).

The results of hematologic tests also indicate that the baby had glucose-6-phosphate dehydrogenase (G6PD) deficiency. Due to the financial disability of the parent to provide his drug and look after the baby, genetic map was not provided and after completing the treatment and make the baby to a stable condition, the baby referred to a rehabilitation center for maintenance with accomplished coordination.

DISCUSSION

The most common defects in the lower limbs are Hemimelia fibular, Hemimelia tibial and Hemimelia femoral (6). In this case, the neonate has aplasia of the lower limb bones, malformation as the absence of tibia, fibula, and the absence of ankle bones.

The fibular hemimelia (FH), the most common congenital anomaly in long bones. In men, it is approximately twice as likely as women. In two-third of cases, it occurs unilaterally, and hereditary transmission does not. With surgery, you can easily treat all types of FH. The cause of this defect remains unclear and is suspected to be due to a disturbance in the molecular signaling of this syndrome (13, 14).

Tibial Hemimelia (TH) is also a common congenital role that occurs in the lower extremity. In traditional societies, it is amputation.TH can be one-way or two-way, approximately 30% one-way. A study also showed that 72% of the THs occur side by side on the right.TH can be associated with many syndromes such as werner syndrome, CHARGE syndrome, and gollop-wolfgang syndrome. Surgery is the best way to treat it. It is believed that the cause of such anomaly is a disturbance in signaling and mutation in the gene, which causes a defect in the protection process. Parents' family relationship is one of the main factors that can lead to these types of abnormalities (15, 16).

This newborn had a deformity in the skull area that was diagnosed plagiocephaly by MRI. Plagiocephaly is a congenital anomaly in the infant, which means asymmetrical deformity in the head and skull (17). It is usually benign, but can sometimes cause some neuropathology disorders. The disease is divided into two types of synostotic and non-synostotic. This newborn had a deformity in the skull area that was diagnosed plagiocephaly by MRI.

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and non-synostotic

Synostotic plagiocephaly is caused due to early closure of the skull sutures, which causes the head to become asymmetrical. Non-synostotic plagiocephaly also happens when the pressure increased and for environmental factors (18). This defect can be a risk factor for other disorders, such as growth retardation and can continue to 36 months of age. It can be treat by orthotic molding helmets, specialized pillows and other therapies such as surgery. These anomalies are typically recognizable in visual examinations (19).

Congenital syndactyly is associated with a wide spectrum of local anomalies. Some cases of syndactyly occur sporadically that no identifiable genetic reason. In some cases, the condition occurs as an inherited trait, and in some cases, syndactyly is an accompanying defect in a genetic syndrome, such as Poland syndrome, Apert syndrome or Holt-Oram syndrome (20). Our patient has several rare anomalies, which can have many problems for him.

CONCLUSION

Congenital anomalies are the cause of many infants' deaths. There are many factors that cause these types of anomalies and There are many ways to prevent these diseases. To detect most of these types of anomalies, MRI, radiography, ultrasound, and computed tomography (CT) scan can be used. Due to the rarity of this condition, such case reports are very infrequent in the literature.

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AUTHOR CONTRIBUTIONS

Mohammad zahedi, Faeze Azadi; Contributed to conception and design. Faeze Azadi, Mohammad Moein Maddah; Contributed to all experimental work and interpretation of data. Mohammad zahedi, Mohsen Hosseinzadegan; Drafted the manuscript, which was revised by Ali Mirabi, Marzieh Zamaniyan. All authors read and approved the final manuscript.

CONFLICT OF INTERESTS

Authors declare no conflicts of interest.

ETHICAL STANDARDS

None.

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